



CUL7 gene

cullin 7

Normal Function

The *CUL7* gene provides instructions for making a protein called cullin-7. This protein plays a role in the cell machinery that breaks down (degrades) unwanted proteins, called the ubiquitin-proteasome system.

Cullin-7 helps to assemble a complex known as an E3 ubiquitin ligase. This complex tags damaged and excess proteins with molecules called ubiquitin. Ubiquitin serves as a signal to specialized cell structures known as proteasomes, which attach (bind) to the tagged proteins and degrade them. The ubiquitin-proteasome system acts as the cell's quality control system by disposing of damaged, misshapen, and excess proteins. This system also regulates the level of proteins involved in several critical cell activities such as the timing of cell division and growth.

Health Conditions Related to Genetic Changes

3-M syndrome

At least 25 mutations in the *CUL7* gene have been identified in people with 3-M syndrome. Some of these mutations substitute one protein building block (amino acid) for another amino acid in the cullin-7 protein. Others result in a cullin-7 protein that is abnormally short and nonfunctional.

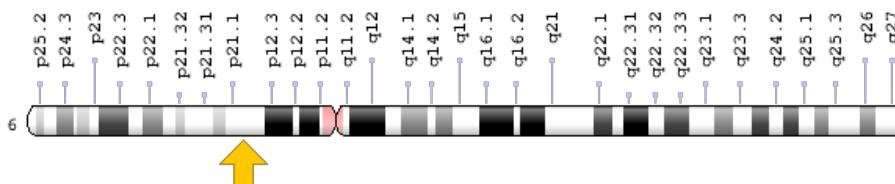
Individuals in the Yakut population of Siberia with a variant of 3-M syndrome all have a particular mutation in both copies of the *CUL7* gene in each cell. This mutation replaces the amino acid glutamine with a premature stop signal in the instructions for making the cullin-7 protein (written as Gln1553Ter or Q1553X).

Mutations in the *CUL7* gene disrupt the ability of the cullin-7 protein to bring together the components of the E3 ubiquitin ligase complex, interfering with the process of tagging other proteins with ubiquitin (ubiquitination). It is not known how impaired ubiquitination results in growth retardation and the other signs and symptoms of 3-M syndrome.

Chromosomal Location

Cytogenetic Location: 6p21.1, which is the short (p) arm of chromosome 6 at position 21.1

Molecular Location: base pairs 43,037,617 to 43,053,950 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CUL7_HUMAN
- dJ20C7.5
- KIAA0076

Additional Information & Resources

GeneReviews

- 3-M Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1481>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CUL7%5BTIAB%5D%29+OR+%28cullin+7%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- CULLIN 7
<http://omim.org/entry/609577>

Research Resources

- **Atlas of Genetics and Cytogenetics in Oncology and Haematology**
http://atlasgeneticsoncology.org/Genes/GC_CUL7.html
- **ClinVar**
<https://www.ncbi.nlm.nih.gov/clinvar?term=CUL7%5Bgene%5D>
- **HGNC Gene Family: Cullins**
<http://www.genenames.org/cgi-bin/genefamilies/set/1032>
- **HGNC Gene Symbol Report**
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=21024
- **NCBI Gene**
<https://www.ncbi.nlm.nih.gov/gene/9820>
- **UniProt**
<http://www.uniprot.org/uniprot/Q14999>

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